



hereditary sensory and autonomic neuropathy type V

Hereditary sensory and autonomic neuropathy type V (HSAN5) is a condition that primarily affects the sensory nerve cells (sensory neurons), which transmit information about sensations such as pain, temperature, and touch. These sensations are impaired in people with HSAN5.

The signs and symptoms of HSAN5 appear early, usually at birth or during infancy. People with HSAN5 lose the ability to feel pain, heat, and cold. Deep pain perception, the feeling of pain from injuries to bones, ligaments, or muscles, is especially affected in people with HSAN5. Because of the inability to feel deep pain, affected individuals suffer repeated severe injuries such as bone fractures and joint injuries that go unnoticed. Repeated trauma can lead to a condition called Charcot joints, in which the bones and tissue surrounding joints are destroyed.

Frequency

HSAN5 is very rare. Only a few people with the condition have been identified.

Genetic Changes

Mutations in the *NGF* gene cause HSAN5. The *NGF* gene provides instructions for making a protein called nerve growth factor beta (NGF β) that is important in the development and survival of nerve cells (neurons), including sensory neurons. The NGF β protein functions by attaching (binding) to its receptors, which are found on the surface of neurons. Binding of the NGF β protein to its receptor transmits signals to the cell to grow and to mature and take on specialized functions (differentiate). This binding also blocks signals in the cell that initiate the process of self-destruction (apoptosis).

Additionally, NGF β signaling plays a role in pain sensation. Mutation of the *NGF* gene leads to the production of a protein that cannot bind to the receptor and does not transmit signals properly. Without the proper signaling, sensory neurons die and pain sensation is altered, resulting in the inability of people with HSAN5 to feel pain.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- congenital insensitivity to pain
- congenital sensory neuropathy with selective loss of small myelinated fibers
- hereditary sensory and autonomic neuropathy, type 5
- HSAN type V
- HSAN V
- HSAN5

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Congenital sensory neuropathy with selective loss of small myelinated fibers
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0020075/>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Degenerative Nerve Diseases
<https://medlineplus.gov/degenerativenervediseases.html>
- Health Topic: Peripheral Nerve Disorders
<https://medlineplus.gov/peripheralnervedisorders.html>

Genetic and Rare Diseases Information Center

- Hereditary sensory and autonomic neuropathy type V
<https://rarediseases.info.nih.gov/diseases/12328/hereditary-sensory-and-autonomic-neuropathy-type-v>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Peripheral Neuropathy
<https://www.ninds.nih.gov/Disorders/All-Disorders/Peripheral-Neuropathy-Information-Page>
- National Institutes of Health Rare Diseases Clinical Research Network: The Inherited Neuropathies Consortium
<http://www.rarediseasesnetwork.org/cms/inc/Healthcare-Professionals/CMT>

Educational Resources

- Disease InfoSearch: Congenital sensory neuropathy with selective loss of small myelinated fibers
<http://www.diseaseinfosearch.org/Congenital+sensory+neuropathy+with+selective+loss+of+small+myelinated+fibers/8080>
- National Institute of Neurological Disorders and Stroke: Hereditary Neuropathies
<https://www.ninds.nih.gov/Disorders/All-Disorders/Hereditary-Neuropathies-Information-Page>
- Orphanet: Hereditary sensory and autonomic neuropathy type 5
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=64752
- University of Chicago Center for Peripheral Neuropathy
<http://peripheralneuropathycenter.uchicago.edu/>

Patient Support and Advocacy Resources

- The Foundation for Peripheral Neuropathy
<https://www.foundationforpn.org/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28hereditary+sensory+and+autonomic+neuropathy+type+v%5BTIAB%5D%29+OR+%28hsan+type+v%5BTIAB%5D%29+OR+%28hsan+v%5BTIAB%5D%29+OR+%28hsan%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE V
<http://omim.org/entry/608654>

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